Annual Report 2005
Protocol #0104-465; Grant 1 RO1 HL069877; and BB-IND 11385
Phase I Trial of Intramuscular Injection of a Recombinant Adeno-Associated Virus
Alpha 1-Antitrypsin (rAAV2-CB-hAAT) Gene Vector to AAT-Deficient Adults

Non-Technical Abstract

Alpha 1-antitrypsin (AAT) deficiency is a genetic disorder resulting in a low level or lack of the protein called AAT. AAT protects the lungs from substances in white blood cells that can cause damage to the air sacs in the lungs. AAT deficiency may result in serious lung disease called emphysema. It may also cause liver disease. Gene transfer is an experimental (investigational) procedure that is being developed to help treat genetic diseases, including AAT deficiency, by putting normal copies of genes into body cells. An investigational, procedure is one that is being tested for safety and has not been approved by the U.S. Food and drug Administration (FDA). Genes are located inside cells, and they cannot be introduced inside the cell without help. In this case, help is in the form of a modified virus called adeno-associated virus (AAV). This is a very small and simple virus that has been genetically engineered in the laboratory to contain a normal copy of the AAT gene. The AAV virus is not known to cause any disease or illness in the human body. When AAV is combined with the AAT gene, the resulting vector (study agent) is called rAAV2-CB-hAAT. This vector can carry normal copies of the AAT gene into the cells of muscle to produce and secrete AAT into the bloodstream. In this study, this is done by injecting rAAV2-CB-hAAT with a needle and syringe into the muscle of the upper arm. The purpose of this study is to test the safety of injecting the rAAV2-CB-hAAT into muscle. This is the first time this study agent has ever been tested in human subjects. 12 subjects are participating in this study.